

Myelodysplastic syndrome with cryptic 5q deletions in young male patients showing sustained response to lenalidomide

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Supplementary data

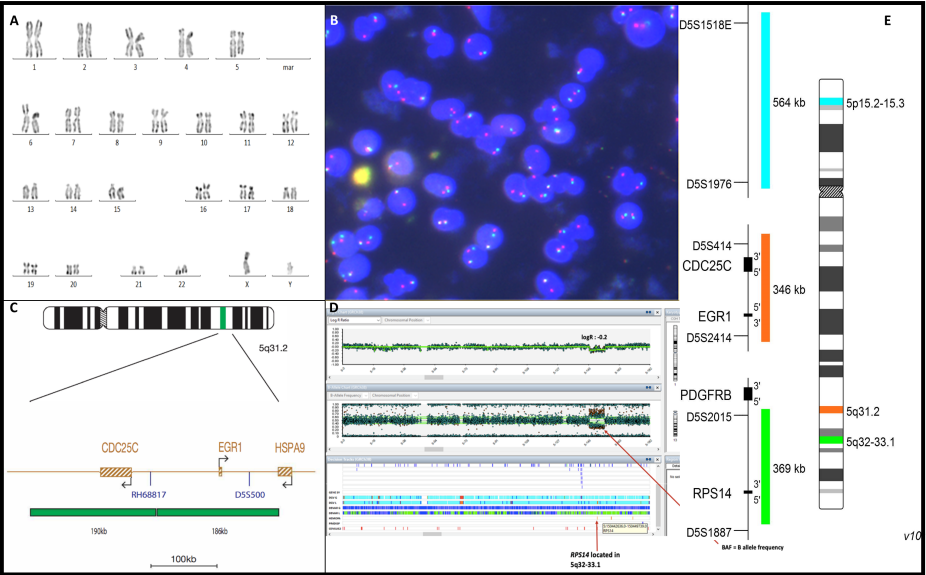


Figure S1 - Cytogenetic results of patient n°1

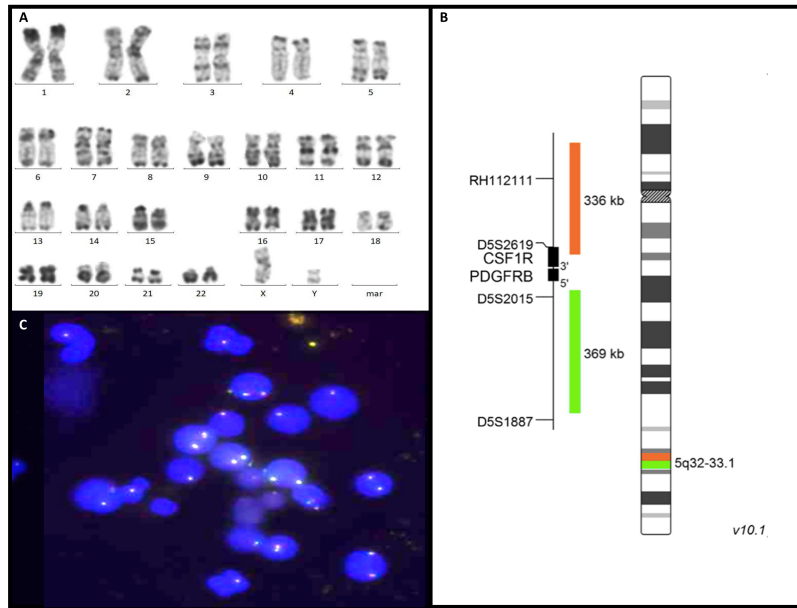


Figure S2 - Cytogenetic results of patient n°2

Reference	Sex	Age at the diagnosis (years)	Blood count	Marrow dysplasia	Deleted Region (size)	Main Gene Deleted	Detection Method	Treatment	Duration of Response
Vlachos et al. (2013) ¹²	F	17	Hb: 9.2 g/dL MCV: 112.4 fL WBC: 16.4G/L Platelet: 478 G/L	Erythroid hypoplasia, increased megakaryocyte number with mild atypia, rare hypolobulated nuclei	5q33 – mosaic deletion in 5q33 (897 kb)	<i>RPS14</i>	SNP array	Not reported	Not reported
Hemmat et al. (2014) ¹³	F	88	Hb: 10.5 g/dL MCV: WBC: 16.2 G/L Platelets: 79 G/L	Occasional atypical granulocytes and megakaryocytes	5q31.2 (896 kb)	<i>CTNNA1</i> <i>HSPA9</i>	SNP array	Not reported	Not reported
Medlock et al. (2017) ¹⁴	F	52	Hb: 9.1 g/dL MCV: 110 fL WBC: 4.35 G/L Platelets: 1701 G/L	Erythroid hypoplasia and a striking increase in non-lobated megakaryocytes	5q33	<i>RPS14</i>	SNP array	Not reported	Not reported
Patient n°1	M	40	Hb: 8.7 g/dL MCV: 112 fL WBC: 3.6 G/L Platelets: 286 G/L	Hypoplastic marrow with multilineage dysplasia. Very rare monolobulated megakaryocytes.	5q32-33.2 (9.3Mb)	<i>RPS14</i>	SNP array	Lenalidomide	26+ months
Patient n°2	M	25	Hb: 9.5 g/dL MCV: 114 fL WBC: 5.02 G/L Platelets: 344 G/L	Monolobated megakaryocytic dysplasia without fibrosis or increased blasts	5q32	<i>PDGFRB</i>	FISH	Lenalidomide	83+ months

Table S1 – Cases of MDS with 5q Microdeletion: Three Literature-Reported Cases and the Two Present Cases

Figures and table legends:

Online Supplementary Figure 1: Cytogenetic results of patient n°1 (A) G-Banded Karyotype of patient n°1 with the suspicion of a small deletion in 5q31 (B) Interphase FISH using CytoCell 5q31.2 FISH probe. It shows 2 green and 2 red spots, concluding to the absence of deletion of *EGR1* and *CDC25C*. (C) CytoCell 5q31.2 FISH probe used on the first karyotyping. This probe targets *EGR1* and *CDC25C*. (D) Capture of the results on DNA sample from bone marrow provided by SNParray NextSeq550 Illumina. It represents the deletion of *RPS14* in 5q31-33.1. LogR ratio that shows the intensity signals of the SNPs indicates a loss in this case (Log R<0). B allele frequency (BAF) shows loss of heterozygotes. (E) XL 5q31/5q33/5p15 Deletion Probe from Metasystems used on the second karyotyping to confirm the deletion of *RPS14* visualized on SNP array. This probe targets *EGR1* and *CDC25C* (orange zone) in 5q31.2 but also *RPS14* (green zone) in 5q32-33.1.

Online Supplementary Figure 2: Cytogenetic results of patient n°2 (A) G-Banded Karyotype of patient n°2 with the suspicion of a microdeletion in 5q32 (B) XL 5q32 *PDGFRB* BA probe from Metasystems used on the second patient's karyotyping. This probe targets specifically *PDGFRB* in 5q32. (C) Interphase FISH using XL 5q32 *PDGFRB* BA probe. It shows 2 green-orange colocalization (yellow) spots in some nuclei (normal) and 1 spot in other nuclei (pathological), concluding to the deletion of *PDGFRB*.

Online Supplementary Table 1: Cases of MDS with 5q Microdeletion: Three Literature-Reported Cases and the Two Present Cases: Key clinical, hematological, and cytogenetic characteristics of three reported cases of 5q microdeletion from the literature, compared with the two patients presented in this study. Parameters include sex, age at diagnosis, hematologic indices (hemoglobin [Hb], mean corpuscular volume [MCV], white blood cell [WBC] count, and platelet count), bone marrow cytology, specific deleted chromosomal regions, key genes affected, detection methods (SNP array or FISH), treatment administered, and duration of clinical response.